Opinion

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# Prenatal genetic screening and the evolving quest for "perfect babies": at what cost for genetic diversity?

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Nearly two decades have passed since the first draft sequences of the human genome were published at the eyewatering cost of nearly US$3 billion for the publicly funded project. Sequencing costs have dropped drastically since, and a range of direct-to-consumer genetics companies now offer partial sequencing of your individual genome in the US$100 price range, and whole-genome sequencing for less than US$1,000.

While such tests are mainly for personal peruse, there have also been substantial drops in price in clinical genome sequencing, which has greatly enabled the study of and screening for inheritable disorders. This has both advanced our understanding of these diseases in general, and benefitted early diagnosis of many genetic disorders, which is crucial for early and efficient treatment. Such detection can, in fact, now occur long before birth: from cell-free DNA testing during the first trimester of pregnancy, to genetic testing of embryos generated by *in vitro* fertilization, to preconception carrier screening of parents to find out if both are carriers of an autosomal recessive condition. While such prenatal testing of foetuses or embryos primarily focuses on diseases caused by chromosomal abnormalities, technological advances allow also for the testing of an increasing number of heritable monogenic conditions in cases where the disease-causing variants are known.

The medical benefits of such screening are obvious: I personally have lost two pregnancies, one to Turner's syndrome and the other to an extremely rare and lethal autosomal recessive skeletal dysplasia, and I know first-hand the heartbreak and devastation involved in finding out that you will lose the child you already love so much. It should be noted though that, very rarely, Turner syndrome is survivable and the long-term outlook is typically good in those cases (GARD, 2021). In addition, I have Kallmann syndrome, a highly genetically complex dominant endocrine disorder (Maoine *et al*, 2018), and early detection and treatment make a difference in outcome. Being able to screen early during pregnancy or childhood therefore has significant benefits for affected children. Many other genetic disorders similarly benefit from prenatal screening and detection.

But there is also obvious cause for concern: the concept of "designer babies" selected for sex, physical features, or other apparent benefits is well entrenched in our society – and indeed culture – as a product from a dystopian future. Just as a recent example, Philipp Ball, writing for the *Guardian* in 2017, described designer babies as "an ethical horror waiting to happen" (Ball, 2017). In addition, various commercial enterprises hope to capitalize on these screening technologies. Orchid Inc claims that their preconception screening allows you to "… safely and naturally, protect your baby from diseases that run in your family". The fact that this is hugely problematic if not impossible from a technological perspective has already been extensively clarified by Lior Pachter, a computational biologist at Caltech (Pachter, 2021). George Church at Harvard University suggested creating a DNA-based dating app that would effectively prevent people who are both carriers for certain genetic conditions from matching (Flynn, 2019). Richard Dawkins at Oxford University recently commented that "…the decision to deliberately give birth to a Down [syndrome] baby, when you have the choice to abort it early in the pregnancy, might actually be immoral from the point of view of the child's own welfare" (Dawkins, 2021).

These are just a few examples, and as screening technology becomes cheaper, more companies will jump on the bandwagon of perfect "healthy" babies. Conversely, this creates a risk that parents come under pressure to terminate pregnancies with "imperfect babies" as I have experienced myself. What does this mean for people with rare diseases? From my personal moral perspective, the ethics are clear in cases where the pregnancy is clearly not viable. Yet, there are literally thousands of monogenic conditions and even chromosomal abnormalities, not all of which are lethal, and we are making constant strides in treating conditions that were previously considered untreatable. In addition, there is still societal prejudice against people with genetic disorders, and ignorance about how it is to live with a rare disease. In reality, however, all rare disease patients I have encountered are happy to be alive and here, even those whose conditions have significant impact on their quality of life. Many of us also don't like the term "disorder" or "syndrome", as we are so much more than merely a disorder or a syndrome.

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Unfortunately, I also see many parents panic about the results of prenatal testing. Without adequate genetic counselling, they do not understand that their baby's condition may have actually a quite good prognosis without major impact on the quality of life. Following from this, a mere diagnosis of a rare disease – many of which would not even necessarily have been detectable until later in life, if at all – can be enough to make parents consider termination, due to social stigma.

This of course raises the thorny issue of regulation, which range from the USA where there is little to no regulation of such screening technologies (ACOG, 2020), to Sweden where such screening technologies are banned with the exception of specific high-risk/lethal medical conditions both parents are known carriers for (SMER, 2021). As countries come to grips with both the potential and the risks involved in new screening technologies, medical ethics board have approached this issue. And as screening technologies advance, we will need to ask ourselves difficult questions as a society. I know that in the world of "perfect babies" that some of these companies and individuals are trying to promote, I would not exist, nor would my daughter. I have never before had to find myself so often explaining to people that our lives have value, and I do not want to continue having to do so. Like other forms of diversity, genetic diversity is important and makes us richer as a society. As these screening technologies quickly advance and become more widely available, regulation should at least guarantee that screening must involve proper genetic counselling from a trained clinical geneticist so that parents actually understand the implications of the test results. More urgently, we need to address the problem of societal attitudes towards rare diseases, face the prejudice and fear towards patients, and understand that abolishing genetic diversity in a quest for perfect babies would impoverish humanity and make the world a much poorer place.

## Conflict of interest

The author declares that she has no competing interests other than having a rare monogenic disorder herself.

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